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Tetrahydrobiopterin in phenylketonuria

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Stellingen

Behorende bij het proefschrift

Tetrahydrobiopterin in Phenylketonuria. Who can benefit?

1. The BH4 loading test (in non-neonates) should last for at least 48 rather than 24 hours as late responders are worth detecting and should be allowed to be tested for true responsiveness. *(this thesis)*
2. Patients carrying two null-mutations in the phenylalanine hydroxylase gene are no candidates for BH4 treatment. *(this thesis)*
3. BH4 responsiveness can be predicted in a BH4 loading test with a baseline phenylalanine concentration < 400 µmol/l when the test is positive, otherwise a retest with phenylalanine loading can be considered. *(this thesis)*
4. Defining BH4 responsiveness is not clear-cut and should take into account that some patients already have a relatively high protein intake and thereby a ≥ 100% increase in natural protein tolerance is not recommended *(this thesis)*
5. An 8 hour or even 24 hour neonatal BH4 loading test can miss long-term BH4 responsive patients. As a delay in treatment should be avoided, an 24 hour neonatal BH4 loading test should be the maximum length or otherwise the BH4 withdrawal test should be considered. *(this thesis)*
6. BH4 treatment appears to be safe (in non-pregnant PKU patients), adverse events are likely to disappear with reduced therapeutic BH4 doses. *(this thesis)*
7. BH4 treatment is associated with lower prolactin concentrations, suggesting higher dopamine availability, independent of a decrease in phenylalanine concentration in male PKU patients. *(this thesis)*
8. Under current conditions BH4 treatment does not affect brain neurotransmitter concentrations in PKU mice, but it does seem to increase brain serotonin concentrations in wild type mice. *(this thesis)*
9. "Treat the patient and the phenotype, not the genome" *(Charles Scriver)*
10. "Chronisch ziek zijn vraagt om chronisch optimisme" *(Loesje.nl)*
11. "Change favors the prepared mind" *(Pasteur)*
12. "Alleen ga je sneller, maar samen kom je verder" *(Afrikaans spreekwoord)*

Karen Anjema

Groningen, 28 april 2020